MOVEMENT DISORDERS GENE PANEL DG 2.3.x

Gene	Median coverage	% covered >10x	% covered >20x	Associated Phenotype description and OMIM ID
ABCB7	102.7	100%	100%	Anemia, sideroblastic, with ataxia, 301310
ABCD1	62.9	76%	74%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABHD12	55.7	96%	90%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	62.8	100%	96%	Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ADCK3	96.5	100%	95%	Coenzyme Q10 deficiency, primary, 4, 612016
ADCY5	81.3	100%	97%	Dyskinesia, familial, with facial myokymia, 606703
AFG3L2	66.8	95%	89%	Spinocerebellar ataxia 28, 610246 Ataxia, spastic, 5, autosomal recessive, 614487
ALDH3A2	91.3	100%	100%	Sjogren-Larsson syndrome, 270200
ANO10	92.2	100%	98%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	99.4	100%	100%	Dystonia 24, 615034
AP4B1	92	100%	100%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	119	100%	99%	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	96	100%	97%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	62.5	92%	88%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	74.2	97%	89%	Spastic paraplegia 48, autosomal recessive, 613647
APTX	104.2	100%	96%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARSA	88.4	99%	97%	Metachromatic leukodystrophy, 250100
ARX	62.7	83%	79%	Epileptic encephalopathy, early infantile, 1, 308350 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Proud syndrome, 300004 Partington syndrome, 309510 Hydranencephaly with abnormal genitalia, 300215
ASPA	100.7	100%	100%	Canavan disease, 271900
ATCAY	94.1	100%	98%	Ataxia, cerebellar, Cayman type, 601238

ATL1	97.9	98%	94%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATM	104.2	100%	99%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell T-cell prolymphocytic leukemia, somati
ATP13A2	77	98%	93%	Parkinson disease 9, 606693
ATP1A3	105.1	100%	99%	Dystonia-12, 128235 Alternating hemiplegia of childhood 2, 614820
ATP2B3	104.7	99%	99%	Spinocerebellar ataxia, X-linked 1, 302500
ATP7B	116.1	99%	96%	Wilson disease, 277900
B4GALNT1	78.5	95%	88%	Spastic paraplegia 26, autosomal recessive, 609195
BCAP31	68.7	82%	74%	Deafness, dystonia and cerebellar hypomyelination, 300475
BCKDHA	103.7	100%	99%	Maple syrup urine disease, type Ia, 248600
BCKDHB	76.7	100%	87%	Maple syrup urine disease, type Ib, 248600
BSCL2	96	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type V, 600794
C10orf2	127.7	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia, autosomal dominant, 3, 609286
C12orf65	145.3	100%	100%	Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	80.4	100%	99%	Neurodegeneration with brain iron accumulation 4, 614298
CA8	77.1	100%	98%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	71.8	96%	90%	Migraine, familial hemiplegic, 1, 141500 Episodic ataxia, type 2, 108500 Spinocerebellar ataxia 6, 183086 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500
CACNB4	80.1	99%	97%	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 Episodic ataxia, type 5, 613855
CCT5	70	91%	81%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CIZ1	90.7	100%	97%	Dystonia 23, 614860
COASY	115.5	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643

COQ2	60.8	97%	88%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ9	80.5	98%	92%	Coenzyme Q10 deficiency, primary, 5, 614654
CP	75.6	97%	91%	[Hypoceruloplasminemia, hereditary], 604290 Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CSTB	150.5	100%	99%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CYP27A1	107.3	95%	93%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	96.6	100%	95%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	88.7	100%	97%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
DBT	93.8	100%	100%	Maple syrup urine disease, type II, 248600
DCAF17	86.3	100%	97%	Woodhouse-Sakati syndrome, 241080
DCTN1	104.6	100%	96%	Neuropathy, distal hereditary motor, type VIIB, 607641 {Amyotrophic lateral sclerosis, susceptibility to}, 105400 Perry syndrome, 168605
DDC	83.4	100%	97%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	126	99%	98%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	91.9	100%	98%	Spastic paraplegia 54, autosomal recessive, 615033
DLAT	94.2	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	121.3	100%	99%	Dihydrolipoamide dehydrogenase deficiency, 246900
DNMT1	89.7	99%	95%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
EIF2B1	93.1	100%	97%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	81.6	100%	100%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	76.3	100%	97%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	109.6	100%	100%	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	94.4	99%	98%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4G1	101.5	100%	98%	Parkinson disease 18, 614251
ELOVL5	71.8	100%	94%	Spinocerebellar ataxia 38, 615957
ERLIN2	105.9	99%	97%	Spastic paraplegia 18, autosomal recessive, 611225

FA2H	54.1	95%	75%	Spastic paraplegia 35, autosomal recessive, 612319
FAR1	82.4	100%	95%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FBXO7	133.9	100%	100%	Parkinson disease 15, autosomal recessive, 260300
FGF14	105.5	100%	100%	Spinocerebellar ataxia 27, 609307
FLVCR1	85.3	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FTL	81.8	100%	94%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
GALC	87.7	100%	98%	Krabbe disease, 245200
GAN	117.8	100%	100%	Giant axonal neuropathy-1, 256850
GBA	60.9	64%	57%	Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, perinatal lethal, 608013
GBA2	115.1	100%	100%	Spastic paraplegia 46, autosomal recessive, 614409
GCDH	69.9	93%	88%	Glutaricaciduria, type I, 231670
GCH1	100.7	100%	100%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GFAP	74.2	100%	95%	Alexander disease, 203450
GJC2	59.9	96%	84%	Leukodystrophy, hypomyelinating, 2, 608804 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480
GLB1	67.6	99%	95%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GNAL	85.9	100%	100%	Dystonia 25, 615073
GOSR2	86	97%	96%	Epilepsy, progressive myoclonic 6, 614018
GPR56	92.8	100%	98%	Polymicrogyria, bilateral frontoparietal, 606854
GRID2	123	100%	99%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRM1	131.1	100%	98%	Spinocerebellar ataxia, autosomal recessive 13, 614831
HEXB	91.2	100%	100%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HPRT1	80.9	100%	99%	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323

HSPD1	13.8	60%	31%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
ITPR1	97	99%	97%	Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
KCNA1	111.2	100%	100%	Episodic ataxia/myokymia syndrome, 160120
KCNC1	137.1	100%	100%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	60.1	77%	66%	Spinocerebellar ataxia 13, 605259
KCNJ10	153.4	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791
KCTD7	78.9	68%	67%	Epilepsy,progressive myoclonic 3, with or without intracellular inclusions, 611726
KIAA0196	87	99%	97%	Spastic paraplegia 8, autosomal dominant, 603563
KIAA0226	86.8	99%	97%	?Spinocerebellar ataxia, autosomal recessive 15
KIF1A	66.5	99%	93%	Spastic paraplegia 30, autosomal recessive, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255
KIF1C	102.2	99%	95%	Spastic ataxia 2,autosomal recessive, 611302
KIF5A	80	99%	95%	Spastic paraplegia 10, autosomal dominant, 604187
L1CAM	114.2	100%	100%	Hydrocephalus due to aqueductal stenosis, 307000 MASA syndrome, 303350 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial
MARS2	140.5	100%	100%	Spastic ataxia 3, autosomal recessive, 611390
MECP2	155	100%	98%	Rett syndrome, 312750 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, preserved speech variant, 312750 Encephalopathy, neonatal severe, 300673 {Autism susceptibility, X-linked 3}, 300496 Angelman syndrome, 105830
MICU1	80	100%	99%	Myopathy with extrapyramidal signs
MMADHC	56	89%	89%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410

MRE11A	80.1	99%	99%	Ataxia-telangiectasia-like disorder, 604391
MTHFR	89.9	100%	98%	Homocystinuria due to MTHFR deficiency, 236250 {Schizophrenia, susceptibility to}, 181500 {Vascular disease, susceptibility to} {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050
MTPAP	87.9	93%	93%	Ataxia, spastic, 4, 613672
MTTP	91.7	100%	97%	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
NIPA1	92.2	97%	86%	Spastic paraplegia 6, autosomal dominant, 600363
NKX2-1	100.8	100%	99%	Goiter, familial, due to TTF-1 defect (1) Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NOL3	122.3	100%	100%	Myoclonus, familial cortical, 614937
NPC1	79.5	99%	95%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	50.5	100%	100%	Niemann-pick disease, type C2, 607625
NUP62	92.1	100%	98%	Striatonigral degeneration, infantile, 271930
OPA1	110.7	100%	99%	Optic atrophy 1, 165500 {Glaucoma, normal tension, susceptibility to}, 606657 Optic atrophy plus syndrome, 125250
PANK2	98.4	100%	98%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PAX6	84.1	100%	99%	Aniridia, 106210 Peters anomaly, 604229 Cataract with late-onset corneal dystrophy, 106210 Keratitis, 148190 Foveal hyperplasia, 136520 Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550 Coloboma, ocular, 120200 Coloboma of
PDE8B	87.5	100%	99%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	59	100%	98%	Basal ganglia calcification, idiopathic, 5, 615483

PDGFRB	81.9	100%	98%	Basal ganglia calcification, idiopathic, 4, 615007 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550
PDHA1	101.4	100%	99%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	92	100%	100%	Lacticacidemia due to PDX1 deficiency, 245349
PDSS1	89.4	91%	87%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	81	100%	97%	Coenzyme Q10 deficiency, primary, 3, 614652
PDYN	146.6	100%	100%	Spinocerebellar ataxia 23, 610245
PEX10	66.9	92%	89%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX7	85.9	91%	88%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PHYH	78.1	100%	100%	Refsum disease, 266500
PIK3R5	73.5	100%	100%	Ataxia-oculomotor apraxia 3, 615217
PLA2G6	71.7	99%	91%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953
PLP1	72.8	100%	97%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	88.2	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNKD	58.5	99%	94%	Paroxysmal nonkinesigenic dyskinesia, 118800
PNKP	66	100%	96%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures and developmental delay, 613402
PNPLA6	82.4	100%	97%	Spastic paraplegia 39, autosomal recessive, 612020
POLG	81.5	98%	93%	Progressive external ophthalmoplegia, autosomal recessive, 258450 Progressive external ophthalmoplegia, autosomal dominant, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 20
PRKCG	88.1	99%	95%	Spinocerebellar ataxia 14, 605361
PRKRA	107.4	100%	100%	Dystonia 16, 612067
PRRT2	71.8	100%	96%	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066

REEP1	85.6	100%	95%	Spastic paraplegia 31, autosomal dominant, 610250 Neuronopathy, distal hereditary motor, type VB, 614751
RNASEH2A	93.7	99%	92%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	89.5	100%	100%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	144.2	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNF170	101.5	100%	100%	taxia, sensory, 1, autosomal dominant, 608984
RTN2	67.4	96%	91%	Spastic paraplegia 12, autosomal dominant, 604805
SACS	142.4	100%	100%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	107.1	100%	98%	Aicardi-Goutieres syndrome 5, 612952 Chilblain lupus 2, 614415 -3
SCN8A	126.9	100%	99%	Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558
SERAC1	85.6	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETX	130.9	100%	99%	Ataxia-ocular apraxia-2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SGCE	70.1	95%	91%	maternally imprinted Dystonia-11, myoclonic, 159900
SIL1	85.5	100%	100%	Marinesco-Sjogren syndrome, 248800
SLC12A6	81.3	100%	100%	Agenesis of coprus callosum with peripheral neuropathy, 218000
SLC16A2	82.1	100%	94%	Allan-Herndon-Dudley syndrome, 300523
SLC19A3	95.9	100%	99%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A3	102.1	100%	100%	Episodic ataxia, type 6, 612656
SLC20A2	82.2	100%	98%	Basal ganglia calcification, idiopathic, 1, 213600
SLC25A15	95.2	96%	80%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 -3
SLC2A1	86.6	100%	100%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847 Dystonia 9, 601042
SLC30A10	130.9	100%	100%	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280
SLC33A1	89.6	100%	98%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC52A2	113	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707

SLC6A3	76.1	100%	99%	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 613135
SLC9A1	102.4	100%	97%	No OMIM disease phenotype
SMPD1	95.3	96%	92%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SNCA	115	100%	100%	Parkinson disease 4, 605543 Dementia, Lewy body, 127750 Parkinson disease 1, 168601
SNX14	89.9	98%	94%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SPAST	92.7	100%	100%	Spastic paraplegia 4, autosomal dominant, 182601
SPG11	94.6	100%	98%	Spastic paraplegia 11, autosomal recessive, 604360
SPG20	101.1	100%	100%	Troyer syndrome, 275900
SPG21	94.2	100%	100%	Mast syndrome, 248900
SPG7	76.9	95%	87%	Spastic paraplegia 7, autosomal recessive, 607259
SPR	57.8	100%	99%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTBN2	91.3	99%	97%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
STUB1	103.1	100%	100%	Spinocerebellar ataxia, autosomal recessive 16, 615768
SUOX	149.5	100%	100%	Sulfite oxidase deficiency, 272300
SYNE1	95	99%	97%	Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
TAF1	127	100%	100%	Dystonia-Parkinsonism, X-linked, 314250
TDP1	98.2	100%	100%	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TECPR2	104.6	100%	99%	Spastic paraplegia 49, autosomal recessive, 615031
TGM6	62.1	94%	85%	Spinocerebellar ataxia 35, 613908
TH	93.4	99%	90%	Segawa syndrome, recessive, 605407
THAP1	98.4	100%	100%	Dystonia 6, torsion, 602629
TIMM8A	53.4	99%	79%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TMEM240	109.5	100%	100%	Spinocerebellar ataxia 21, 607454

TMEM67	101.3	100%	100%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TOR1A	112.4	100%	99%	Dystonia-1, torsion, 128100 Dystonia, early-onset atypical, with myoclonic features {Dystonia-1, modifier of}
TREX1	127.6	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TTBK2	108.4	100%	100%	Spinocerebellar ataxia 11, 604432
TTC19	61.9	89%	84%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTPA	76.9	100%	89%	Ataxia with isolated vitamin E deficiency, 277460
TUBB4A	57.5	86%	75%	?Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
VAMP1	89.7	99%	87%	spastic ataxia 1, autosomal dominant
VCP	97.5	99%	96%	Spastic Ataxia 1, autosomal dominant, 108600
VLDLR	103	100%	100%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13A	103.6	100%	98%	Choreoacanthocytosis, 200150
VPS37A	67.4	99%	94%	Spastic paraplegia 53, autosomal recessive, 614898
WDR45	82.9	99%	96%	?Neurodegeneration with brain iron acculation 5, 300894
WDR81	110.8	100%	98%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WWOX	93.5	97%	97%	Esophageal squamous cell carcinoma, 133239 Spinocerebellar ataxia 12, 614322
XPR1	88.4	100%	100%	Basal ganglia calcification, idiopathic, 6, 616413
ZFYVE26	85.5	97%	91%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	71.9	100%	96%	Spastic paraplegia 33, autosomal dominant, 610244
ZNF592	104.4	93%	91%	Spinocerebellar ataxia, autosomal recessive 5, 606937

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
Median Coverage describes the average number of reads seen across 50 exomes
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x

OMIM release used for OMIM disease identifiers and descriptions : June 30th, 2015
This list is accurate for all panel versions starting with DG 2.3. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors